MICHIGAN NEWBORN SCREENING

Practioners Manual 2007

Michigan Department of Community Health, Newborn Screening
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Introduction

Newborn screening saves lives and protects the health of Michigan infants. Since 1965, all Michigan newborns have been screened shortly after birth to see if they are at risk for having rare but preventable genetic disorders. If untreated, these disorders can lead to illness, physical disability, mental retardation, or death. Medication and changes in diet can help prevent most health problems caused by disorders detected by newborn screening.

Whether your primary role is as a primary care provider, neonatologist, pediatric or neonatal nurse practitioner, nurse clinician, nurse, laboratory professional, or support staff member, you play an important role in newborn screening. Most primary care providers will, at some point, receive notice of an abnormal newborn screen. NICU staff are much more likely to deal with newborns with abnormal screening results. Nursery staff will be involved in the follow-up of abnormal results, collection of repeat specimens, and assurance that all infants in their units have documented newborn screening results. While the disorders included in the newborn screening panel are individually rare, approximately 150 affected Michigan infants are identified by newborn screening each year. The Provider Manual is intended to be a newborn screening reference tool and contains background information and general guidance on common issues related to newborn screening The manual does not replace medical advice available by contacting appropriate sub-specialists such as those included in appendix -6.

Overview of Michigan Newborn Screening

DRIED BLOOD SPOT SCREENING

Newborn screening is an important, life-saving part of health maintenance for all newborns. Michigan is a leader in newborn screening, and the Michigan Department of Community Health (MDCH) now screens for 49 disorders (see appendix 4). The 50th disorder, cystic fibrosis, will be added in 2007. The Michigan Public Health Code mandates newborn screening. The mandate does not include exceptions for religious beliefs or for other reasons. If parents object to screening, it is suggested that the hospital request that parents sign a waiver stating that they were informed of the risk to their newborn if screening is declined. See appendix 1 for newborn screening legislation.

Before a newborn is discharged from a birthing center, drops of heel-stick blood are collected on a newborn screening card purchased from MDCH. A picture of the current newborn screening card is included in appendix 7.

Ideally, blood specimens should be collected at 24-36 hours of age. The hospital should send specimens to the MDCH Newborn Screening Laboratory daily. Laboratory testing is typically completed within two to three days of specimen receipt, and all newborn screening results are mailed to the hospital/health care provider that submitted the specimen.

The MDCH Newborn Screening Program is unable to perform STAT laboratory testing. If you are caring for a newborn, that has been previously screened and subsequently develops an acute metabolic crisis, it is appropriate to contact the newborn screening program to obtain screening results. However, if a newborn is suspected of having a disorder that is included in the newborn screening panel, it is appropriate to clinically evaluate the newborn rather than assume that screening results will be available with the rapidity required in an emergency situation. Endocrine, metabolic and hematology specialists are accessible for guidance in such circumstances (see appendix 6).

MDCH will notify the submitting hospital and the health care provider identified on the specimen card, if a repeat specimen is required because the initial specimen was borderline positive for a disorder or unsatisfactory for testing. If the newborn has been discharged or transferred, the repeat request should be forwarded to the newborn's current provider.

When a newborn screen is strong positive for a disorder, MDCH will contact the submitting hospital and health care provider by telephone and fax. In addition, appropriate sub-specialists are also notified by telephone and fax to ensure that appropriate diagnosis and treatment occur.

Beginning March 1, 2007, the MDCH Newborn Screening Program will undertake an initiative to improve the screening process for infants with birth weights under 1800 grams. Due to their immaturity, these small infants are more likely to have conditions that are missed by standard screening protocols. Pre-maturity, together with the therapeutic regimens that very low birth weigh infants require, also make false positive results more likely. When an infant weighing less than 1800 grams is born, **in addition to ordering the initial newborn screen, request that specimens be collected for the MDCH Newborn Screening Program at 14 and 30 days of age.** Reviewing the results of the three screens, as the infant matures, is likely to give a more accurate assessment of risk that the newborn has or does not have one of the newborn screening disorders. This reflexive re-screening protocol is likely to reduce both false positive and false negative results for NICU infants.

Specimens from infants weighing less than 1800 grams at birth should be collected on an MDCH blue newborn screening card at 24-36 hours of life. **If the newborn is transfused at less than 24 hours of age, obtain the newborn screen prior to transfusion.** A second specimen should be collected at 14 days and a third specimen at 30 days on MDCH pink cards (see appendix 3).

Look for highlighted text throughout this manual for more information on the initiative to improve newborn screening for infants weighing less than 1800 grams (see appendix 10).

HEARING SCREENING

Approximately 250 deaf and hard of hearing infants are identified, annually, by newborn hearing screening in Michigan. Although hearing screening is not currently mandated, all Michigan infants should have hearing screening as a standard of care. NICU infants are at increased risk for hearing loss when compared to the general newborn population. Hearing screening of premature, ill, and infants with birth defects can be problematic due to confounding factors presented by their conditions and the treatment they require. Michigan has instituted a **mandated reporting system** for universal newborn hearing screening. The first goal of the hospital-based program is to screen all infants by one month of age. Infants who exhibit evidence of hearing loss should have an hearing assessment by an audiologist by three months of age and early intervention services by six months of age. Hearing screening should be completed by one month of age through either of the following methods: Otoacoustic Emissions (OAE) or Automated Auditory Brainstem Response (AABR). The NICU should have a protocol to appropriately screen the hearing of infants. When MDCH is informed about an infant who does not pass the hearing screen, additional information is requested from the hospital and recommendations for referral and follow-up are sent to the family doctor. Please contact the MDCH Early Hearing Detection Intervention (EHDI) Program to receive the “Medical Follow-up Protocol: Newborn Hearing Screening”. For additional information see appendix 5.

Newborn Screening Practice and Procedures

TIMING OF THE SPECIMEN COLLECTION

Specimens should be collected between 24-36 hours of life. If a newborn is to be transfused before 24 hours of age, collect specimen prior to transfusion.

COMPLETING THE NEWBORN SCREENING CARD

It is extremely important to fill out the screening card completely and accurately. The specimen submitter is legally responsible for the accuracy and completeness of the information on the newborn screening card. The card will be scanned into the database so legibility is critical. **Press firmly using a black pen**, and record the following information in the spaces provided.

INFANT INFORMATION:

- ✓ **INFANT'S NAME:** Record last name followed by first name. If no first name is available at the time of specimen collection, the last name followed by "boy" or "girl" should be used. For single mothers, use the last name of mother or last name specified by mother. **DO NOT LEAVE BLANK.**

- ✓ **GENDER:** Completely shade in the appropriate oval to designate newborn's gender as male or female.

- ✓ **BIRTH DATE:** Use a six-digit number (mm/dd/yy) for date of birth. For example, a birth January 4, 2007 would be recorded as 010407.

- ✓ **BIRTH TIME:** Record time of birth in military time. For example, a birth at 4:30 pm would be recorded as 1630. Note: This information is only required on the “blue” first newborn screening card.

- ✓ **BIRTH WEIGHT, GRAMS:** Record the birth weight in **grams** in the boxes provided. **Do not use pounds and ounces.** Note: This information is only required on the “blue” first newborn screening card.

- ✓ **CURRENT WEIGHT, GRAMS:** Record the current weight in **grams** in the boxes provided. **Do not use pounds and ounces.** Note: This information is only required on the “pink” repeat newborn screening card.

- ✓ **GESTATIONAL WEEKS:** Record weeks of gestation at time of birth. Note: This information is only required on the “blue” first newborn screening card.

- ✓
- ✓ **SINGLE BIRTH:** Completely shade in oval for single birth.

- ✓ **MULTIPLE BIRTH ORDER:** Completely shade in oval to record birth order by "A", “B”, “C" etc. for twins, triplets, etc.

- ✓ **SPECIMEN DATE:** Use a six-digit number (mm/dd/yy) representing the date on which the specimen was obtained.

- ✓ **COLLECTION TIME:** Record time of specimen collection in military time.

- ✓ **COLLECTED BY:** Record initials of person collecting the specimen.

- ✓ **NICU / SPECIAL CARE:** Completely shade in oval “no” or “yes” to indicate if the newborn was in an NICU or special care nursery when the specimen was collected.

- ✓ **RBC TRANSFUSION:** Completely shade in oval “no” or “yes” to indicate whether the newborn was ever transfused with red blood cells **prior** to specimen collection. If yes, give date (mm/dd/yy).

- ✓ **MEDICAL RECORD #-NEWBORN:** Record the birth hospital's identification or medical record number.

- ✓ **TPN FEEDING:** Completely shade in oval “yes” if the newborn is receiving total parenteral nutrition (TPN).

- ✓ **ANCESTRY:** Completely shade in oval for Hispanic or Non-Hispanic.

- ✓ **RACE:** Completely shade in oval for race. If the newborn is of mixed race and has one white parent, select the race of the non-white parent. If of mixed race and both parents are non-white, select “multiracial**”.**

MOTHER INFORMATION:

- ✓ **MOTHER'S NAME:** Record last name followed by first name. If the newborn is going to be released at birth to adoptive or foster parents, provide contact information of adoptive or foster mother. Please note, in black ink above mothers name, that contact information is for adoptive or foster mother. Do not place sticky notes on the card or use red ink, neither will be recorded when the card is scanned into the system. If contact information on new parents, foster parents, or the adoption agency is not on the card, we will not be able to contact the family if necessary. We would like to avoid calling the birth mother if she is not longer responsible for the care of the newborn.

- ✓ **MOTHER'S ADDRESS:** Record mother's current street address, followed by city, state and zip code. Information about the mother is needed for to locate newborns in need of clinical evaluation or retesting.

- ✓ **MOTHER'S PHONE:** Record mother's area code and home telephone number.

- ✓ **MOTHER'S SOCIAL SECURITY NUMBER:** Record mother's social security number. This important information is used to match initial results with repeat tests. If the mother has no social security number, enter the word **NONE** in the first four boxes.

- ✓ **MEDICAL RECORD NUMBER-MOTHER'S:** Record the hospital identification or medical record number. Note: This information is only required on the "blue" first newborn screening card sample.

- ✓ **BIRTH DATE:** Record the mother's date of birth (mm/dd/yy).

- ✓ **HEPATITIS B SURFACE ANTIGEN (HBsAg):** Provide date of test (mm/dd/yy) and completely shade in oval appropriate to indicate positive or negative. **If there is no HBsAg test result in the mother's record, the test should be done immediately.** Notify Patrick Fineis, MDCH Hepatitis B Case Manager, of any positive HbsAg result. He can be contacted by telephone at (517) 335-9443 or email fineisp@michigan.gov. This very important information helps assure that infants at risk receive the proper immunizations. Note: This information is only required on the "blue" first newborn screening card.

PHYSICIAN INFORMATION:

- ✓ **PHYSICIAN'S NAME:** Record last name, followed by first name, of the physician or health care provider to be notified of an unsatisfactory or positive newborn screening test. If the mother does not provide a physician's name, the physician in charge of the newborn nursery should be listed on the NBS card. The physician should arrange for all retesting through the hospital's outpatient laboratory. If the newborn is expected to be in the NICU for at least a week, list a staff neonatalogist as the physician, and write the NICU phone and fax numbers on the NBS card. If discharge is expected within a week, write the name and clinic phone number of the provider who will be taking care of the newborn after discharge.

- ✓ **PHYSICIAN'S PHONE:** Provide physician's area code followed by telephone number. **It is very important to provide a complete and correct number.** This information is used to contact the physician or health care provider with positive test results and follow-up information. If the hospital newborn nursery chooses to follow-up positive results directly, provide the name and telephone number of the staff person designated to contact the family. This option is preferred for newborns without a designated primary care provider.

SUBMITTER INFORMATION

- ✓ **SUBMITTER NAME**: Record the name of the submitter (this should be the birth hospital or midwife on all initial newborn screens). If abbreviation of the hospital's name is necessary, use some letters from each word in the hospital's name (for example, the abbreviation for St. Joseph Mercy Hospital would be St. Jos. Mrcy.).

- ✓ **HOSPITAL CODE**: All birthing hospitals have been assigned a 3-digit hospital code that must be recorded in the boxes provided. The 3-digit code should be listed **before** the two preprinted zeros. For regular nurseries an "0" should be added to the last box (after the two preprinted zeros). For the NICU, a "1" should be added to the last box. For the special care nursery, a "2" should be added to the last box.

- ✓ **SUBMITTER ADDRESS**: Record the submitter's street address followed by the city, state and zip code.

- ✓ **SUBMITTER PHONE:** Record submitter's area code and phone number.

- ✓ **BIRTH HOSPITAL:** Record name of birth hospital here **only if different from the submitter.**

SPECIMEN COLLECTION

Direct collection from a heel puncture is preferred for optimal laboratory results. Blood collection using capillary tubes is discouraged because it increases the risk of a layered specimen or a torn or chafed card. If capillary tube collection becomes necessary due to clinical circumstances, use a fresh heparinized tube for each circle to be filled (EDTA is a coagulant and may interfere with analysis). Touch the tip of the capillary tube to the blood drop from the heel and allow the blood to flow into the tube. The tube may fill better by holding it in a near horizontal position as it touches the drop of blood. Immediately after filling the capillary tube, apply the contents to the center of the first circle on the newborn screening card, allowing the blood to flow out and fill the circle. Waiting too long may allow the blood and plasma to separate and interfere with test analysis. Do not touch the tube to the filter paper. Do not daub the blood on or "fill in" the circle. These actions can result in an unsatisfactory specimen because of scratching or compressing the paper or layering and over-filling the circle. For more information on collection of blood for newborn screening see appendix 2 and NCCLS Blood Collection on Filter Paper for Newborn Screening Programs; Approved Standard – Fourth Edition, Vol 23, No. 21LA4-A4, available on request from the Newborn Screening Program.

Special Circumstances: Early Specimens

Specimens collected at less than 24 hours of age are considered early specimens. Most of the newborn screening disorders are detected by increases in the level of specific analytes (amino acids, fatty acids organic acids, TSH, 17-OHP, detection of an abnormal protein (hemoglobinopathies) or decreased enzyme activity (galactose transferase, biotinidase). A few are detected by decreases in a specific analyte (low carnitine, low T4). Cut offs for disorders detected by elevations or decreases in specific analytes are determined for specimens obtained at greater than or equal to 24 hours of age. Therefore, screening results on specimens obtained prior to 24 hours are inconclusive. All but three of the disorders are screened by tandem mass spectrometry. The hemoglobinopathies are screened by HPLC (High Performance Liquid Chromatography) and two disorders, biotinidase deficiency and Galactosemia, are screened by enzyme assay. Results for these three screens are valid before 24 hours. Collect a specimen before 24 hours of age in the following clinical circumstances:

(1) If an infant requires any type of blood transfusion in the first 24 hours of life, a newborn screening specimen should be collected before transfusion.

(2) If an infant requires surgery in the first 24 hours of life, a newborn screening specimen should be collected pre-operatively.

(3) If an infant requires ECMO (Extracorporeal Membrane Oxygenation) in the first 24 hours of life, a newborn screening specimen should be collected before the pump is started.

(4) If a newborn is unlikely to survive the first 24 hours of life, the newborn screening specimen should be collected.

(5) If a newborn is discharged from the hospital prior to 24 hours of age, a newborn screening specimen should be collected and arrangements should be made to collect a subsequent specimen.

Interpreting screening results for specimens obtained from newborns less than 24 hours of age is difficult, and both false positive and false negative results are more likely to occur. A repeat specimen should be obtained on all newborns screened before 24 hours of age. All positive screening results from early specimens are reported to the submitting physician with appropriate instructions for follow-up. Hemoglobinopathies, galactosemia and biotinidase deficiency, and screening results are valid on early specimens. All other results, positive or negative, cannot be accurately interpreted.

For specimens collected before 24 hours of age from infants weighing less than 1800 grams, MDCH will include a different request for repeat specimens on the infant's report. The comment will read:

"The information provided indicates that the infant was less than 24 hours of age when the specimen was collected. For specimens obtained prior to 24 hours of age, the results of newborn screening are not valid for amino acid disorders, fatty acid oxidation disorders, organic acid disorders, and endocrine disorders. The results are valid for galactosemia, biotinidase deficiency and hemoglobins. Please follow the NICU under 1800 protocol and send a repeat specimen at 14 days of life and a second repeat specimen at 30 days of life."

Special Circumstances: Transfusion

(1) An infant who requires a transfusion before 24 hours of age should have a newborn screening specimen collected prior to transfusion.

(2) If an infant is transfused before the newborn screening specimen is obtained note the time and date of transfusion on the newborn screening card. Screening of the <1800 gram newborn transfused prior to obtaining the newborn screen is no different than screening the >= 1800 gram transfused baby. In either case obtain a repeat specimen >72 hours after the last transfusion date and an additional repeat specimen for hemoglobins 90-days after the last transfusion date. If the newborn leaves the hospital prior to the 72 hour or three-month intervals, include instructions for collection of the repeat newborn screen in the discharge summary to the primary care provider.

Special Circumstances: Transferred Infants

(1) In Michigan, the birth hospital is responsible for ensuring that newborn screening specimens are collected and sent to MDCH. The birth hospital should notify the receiving facility of the newborn screening status. The transferring hospital should include verification that the newborn was or was not screened in the transport paperwork and the receiving hospital should verify the screening status of all transferred newborns. If screening cannot be verified, the receiving hospital should obtain the newborn screen.

(2) If you are transferring an infant who is less than 24 hours old and has not been transfused, but is so unstable that the infant may expire or require a transfusion upon arrival at the next facility, a newborn screening specimen should be collected prior to transfer.

(3) Each state has different newborn screening policy. If you admit an infant born in another state, you should try to obtain the newborn's screening status from the birth facility. If screening status cannot be verified, collect a newborn screen. If a Michigan newborn is transferred to another state, a newborn screen should be obtained prior to transfer.

Special Circumstances: Newborn at high risk of having a newborn screening disorder

The newborn screening program should be notified immediately if a newborn or a newborn's sibling is suspected of having a newborn screening disorder. A sub-specialist will be contacted and provide recommendations on clinical management prior to diagnostic confirmation.

Special Circumstances: Newborn death or pending death.

The newborn screening program should be notified if a newborn dies or is expected to die. A newborn screening specimen should be collected to determine if the newborn has a newborn screening disorder. This information is important for parents in planning future pregnancies. For expected deaths, notify the newborn screening program when death occurs. This will prevent unnecessary notification of parents regarding subsequent screening or diagnostic testing.

False Negative and False Positive Results:

All screening methodologies are subject to false positive and false negative results. The laboratory and clinicians must work together to ensure that true cases are not missed while the number of false positives is minimized. Because of the serious clinical consequences when disorders on the newborn screening panel are untreated, avoiding false negatives is crucial. Normal ranges for newborn screening tests are generally based on the population as a whole rather than on premature and ill newborns found in the NICU. The MDCH Newborn Screening Program works to reduce both false positive and false negative results in this special population. However, given their clinical state and the modalities needed to treat them, false positive and false negative results are more common in pre-term infants.

False negative results (causing delayed treatment of affected infants) have been documented in NICU infants for the following disorders:

- Amino acidopathy (infant on TPN, no post-TPN specimen submitted).
- Congenital adrenal hyperplasia (infant treated with steroids at delivery or mom was administered steroids prior to delivery).
- Galactosemia (specimen submitted post-transfusion, no repeat specimen submitted)
- Congenital hypothyroidism (delayed TSH elevation in pre-term newborn).
- Sickle cell disease (specimen collected after transfusion, no repeat specimen submitted).

The initiative to reflexively re-screen (at 14 and 30 days) infants weighing less than 1800 grams at the time of birth is designed to reduce false negative results. The repeat screens should be helpful in finding cases where thyroid disease is missed due to delayed TSH elevation and where congenital adrenal hyperplasia is not revealed because of treatment with steroids either pre-delivery or post-delivery.

False positive results are also more common in the NICU and are most likely to be caused by the physiologic immaturity of these infants and the side effects of required treatment regimens. False positive results have been seen in NICU infants for the following disorders:

- Amino acidopathy (TPN is seen as an atypical pattern on tandem mass spectroscopy results, as is nonspecific liver disease seen more commonly in premature infants).
- Biotinidase deficiency (TPN, antibiotics, and jaundice have been associated with reduced enzyme activity)
- Congenital adrenal hyperplasia (premature infants have a different adrenal response than full-term infants, also seen most commonly when collected before 24 hours).
- Congenital hypothyroidism (low T4 in pre-term newborns in screening programs with a primary T4 screening algorithm).

The initiative to reflexively re-screen infants weighing less than 1800 grams at the time of birth is designed to reduce multiple, false positive results. If the initial congenital adrenal hyperplasia screen in an infant weighting less than 1800 grams is positive, an immediate retest is not required, follow the same 14 and 30 day retesting sequence as with other newborns <1800 grams. Clinical evaluation is recommended. If the infant has ambiguous genitalia, abnormal electrolytes, or other clinical signs and symptoms consistent with congenital adrenal hyperplasia, consult a pediatric endocrinologist. If there are abnormal results on the congenital adrenal hyperplasia screen at 14 or 30 days of age follow the same protocol that is followed for infants >= to 1800 grams (collection of serum 17-OHP and consultation with a pediatric endocrinologist).
The newborn screening program is currently evaluating a 2nd tier test for CAH. Initial results indicate a 90% reduction in the CAH false positive rate and confirmation of all true positive detected by the current screening protocol. The 2nd tier protocol is scheduled for implementation on January 1, 2008.

Health Care Provider Responsibilities in Newborn Screening

FOLLOW-UP OF POSITIVE NEWBORN SCREENING RESULTS

When MDCH identifies a strong positive result, the physician/health care provider is immediately notified by phone and fax. MDCH will include the following items in the fax notification:

- Newborn screening results
- Action required
- Sub-specialist contact information

Simultaneously, MDCH notifies the appropriate sub-specialist. The health care provider will be contacted by the consulting sub-specialist to develop a plan of action for necessary diagnostic testing and evaluation that is congruent with clinical status.

The MDCH newborn screening program may ask the health care provider or hospital for additional information over time as part of program evaluation and long-term follow-up. The requests for information are required for newborn screening follow-up and are not subject to limitations of the Health Information Portability and Accountability Act (HIPAA). See appendix 8 for an explanation of why information pertaining to follow-up of abnormal newborn screening results is exempt from HIPAA.

DOCUMENTATION OF NEWBORN SCREENING RESULTS

Documentation that a newborn has been screened should be available for every newborn and included in the medical record. Tracking repeat specimens (because initial specimen was borderline positive, collected before 24 hours of age, post-transfusion or unsatisfactory for testing) is important. The provider is responsible for facilitating subsequent testing.

For infants weighing less than 1800 grams at birth, the provider should order and then ensure that repeat screens are collected at 14 and 30 days of age. If the infant is ready for discharge before a subsequent screen is due to be collected, collect that final specimen on the day of discharge.

Do not assume that no news is always good news. If you cannot locate newborn screening results, please verify that screening was done. If results are not received within two weeks following sample submission, first contact your hospital laboratory and/or medical record department for results or contact the hospital of birth for newborns transferred to your hospital. If no report can be found, contact the MDCH Newborn Screening Program to obtain a copy of the results. It may be helpful to

check to see if the newborn was known by a different name at the time of initial screening.

Include newborn screening results in all NICU discharge summaries. If results are not yet available, include information on date of collection and instructions for the primary care provider to obtain results from the birth hospital medical records department and if not available there, contact MDCH for a copy.

For infants born weighing less than 1800 grams, include the results of all screens in the discharge summary. If a result is still pending, be certain to include that fact since primary care providers are likely to be unfamiliar with the repeat screening algorithm for very small infants.

If a repeat specimen is required because a transfusion was administered prior to collection of the first sample, the 90-day interval for collecting a final specimen (for hemoglobins) should be calculated from the date of the last transfusion. If the newborn leaves the hospital prior to this interval, include instructions for collecting the repeat newborn screen in the discharge summary to the primary care provider.

PARENTAL REFUSAL OF NEWBORN SCREENING

There is no provision in the Michigan newborn screening law to opt out of screening. However, **if parents strongly object to newborn screening**, they should be asked to sign a document that indicates that they have been informed of the risk to their newborn if screening is not done. A copy of the signed document should be forwarded to the MDCH Newborn Screening Program (see appendix 9).

Frequently Asked Questions:

Who informs parents about newborn screening?

The birth hospital is ultimately responsible for informing parents about the newborn screening process. Education is ideally done during the prenatal period. Many families with infants in the NICU will be unfamiliar with newborn screening. Early deliveries and more urgent matters often preclude discussions about this important part of infant health care. To facilitate talking with parents, the MDCH Newborn Screening Program recommends using the newborn screening parent brochure as a tool.

What is the chance that an infant will have a disorder detected by newborn screening?

The chance that an infant will have one of the disorders detected by newborn screening is small. Each year, approximately one in 800 Michigan infants (about 150 infants) will be diagnosed with one of the disorders. In these cases, early diagnosis and treatment may prevent many adverse outcomes associated with these conditions. Health care providers should treat all abnormal screening results urgently and complete recommended follow-up testing or evaluations without delay.

What if an infant has a family history of a disorder detected by newborn screening?

In addition to newborn screening, infants who have a family history of a disorder detected by newborn screening should have definitive diagnostic testing for that particular disorder. This additional diagnostic testing after birth is necessary even if prenatal testing was performed. Please inform the MDCH Newborn Screening Program if a family has a history of a disorder on the newborn screening panel by completing in the “Other” space on the newborn screening specimen card.

What is the Newborn Screening Program’s specimen storage policy?

Newborn screening specimens are stored securely for 21.5 years. MDCH may use specimens in the laboratory for quality control purposes or new test development. Researchers outside MDCH may use specimens only if the specimens cannot be traced to individuals and the research has been approved by the department’s Institutional Review Board.

Who decides which disorders are included on the newborn screening panel?

Based on nationally accepted criteria, the Michigan Department of Community Health Newborn Screening Quality Assurance Advisory Committee makes recommendations on disorder inclusion to the Director of the Michigan Department of Community Health. The Newborn Screening Quality Assurance Advisory

Committee meets once each year to discuss newborn screening issues. Members include parents of affected children, health care providers, hospital representatives, and other medical experts

What if I need to talk to someone at the MDCH Newborn Screening Program or a specialist?

Call 1-866-673-9939 to reach someone in the newborn screening program or see Appendix 6 to contact a specialist or Appendix 12 to review other NBS resources and contacts.

Resource List - 2007

Genetics Home Reference:
http://ghr.nlm.nih.gov/

The information that is specific to Newborn Screening is:
http://ghr.nlm.nih.gov/ghr/search?query=metabolic+disorders

Michigan Newborn Screening:
http://www.michigan.gov/newbornscreening

Order form for MI brochures:
http://www.michigan.gov/documents/Order_form_134956_7.pdf

Course on Newborn Screening
http://training.mihealth.org/

National Newborn Screening and Genetics Resource Center
http://genes-r-us.uthscsa.edu/

The March of Dimes has just developed a 5 min DVD to explain Newborn Screening to parents. Michigan March of Dimes will send hospitals a free copy. Spanish and English versions are also available. It would be a great tool if families do not read, plus it is fabulously produced for everyone to understand the information.

Please contact Kara below to request the DVD. She will take fax, phone or e-mail orders. For reference, the video product number is 09-2099-06

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Southfield, MI 48034
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EHDI Hearing Contact Early Hearing Screening Detection and Intervention Division of Family and Community Health 517-335-8955, FAX 517-335-8036 Michigan EHDI Web page http://www.michigan.gov/ EHDI	NBS Hepatitis B Contact Perinatal Hepatitis B Coordinator Division of Immunization 517-335-9443, FAX 517-335-9855

Michigan Newborn Screening Contact Information

Follow-up Office:

Phone (517) 335-9205 or Toll-free (877) 673-9939

Fax: (517) 335-9739 or (517) 335-9419

E-mail: mdch-newbornscreening@michigan.gov

Web site: http://www.michigan.gov/newbornscreening

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Newborn Screening
201 Townsend St
P.O. Box 30195
Lansing, Michigan 48909

State Laboratory:

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Acknowledgements

Michigan would like to acknowledge and thank Minnesota Department of Health staff Abbie Abboud and Beth-Ann Bloom, for their creation of a prototype of this document and willingness to share it for the benefit of Michigan's children. Ouestions regarding their manual can be directed to:

Phone: (800) 664-7772 (651-201-5797 TDD)
Fax: (651) 201-5471

E-mail: newbornscreening@health.state.mn.us
Website: http://www.health.state.mn.us/newbornscreening

Building Address (for visitors, couriers, deliveries):
Minnesota Department of Health
Newborn Screening Program
601 North Robert Street
St. Paul, MN 55155

Mailing Address (for US mail):
Minnesota Department of Health
Newborn Screening Program
P.O. Box 64899
St. Paul, MN 55164-0899

Appendix 1

Legislative Mandates

Public Health Code

The newborn screening program applies to all newborns in the State of Michigan by law. You can find the law in its entirety at the following link:

http://www.legislature.mi.gov/(S(cedw22550qdahg55ceyqja2n))/mileg.aspx?page=getobject&objectname=mcl-333-5431

PUBLIC HEALTH CODE (EXCERPT)
Act 368 of 1978

333.5431 Testing newborn infant for certain conditions; reporting positive test results to parents, guardian, or person in loco parentis; compliance; fee; "Detroit consumer price index" defined; violation as misdemeanor; hardship waiver; conduct of department regarding blood specimens; pamphlet; additional blood specimen for future identification.

Sec. 5431.
(1) A health professional in charge of the care of a newborn infant or, if none, the health professional in charge at the birth of an infant shall administer or cause to be administered to the infant a test for each of the following:
(a) Phenylketonuria.
(b) Galactosemia.
(c) Hypothyroidism.
(d) Maple syrup urine disease.
(e) Biotinidase deficiency.
(f) Sickle cell anemia.
(g) Congenital adrenal hyperplasia.
(h) Medium-chain acyl-coenzyme A dehydrogenase deficiency.
(i) Other treatable but otherwise disabling conditions as designated by the department.
(2) The informed consent requirements of sections 17020 and 17520 do not apply to the tests required under subsection (1). The tests required under subsection (1) shall be administered and reported within a time and under conditions prescribed by the department. The department may require that the tests be performed by the department.
(3) If the results of a test administered under subsection (1) are positive, the results shall be reported to the infant's parents, guardian, or person in loco parentis. A person is in compliance with this subsection if the person makes a good faith effort to report the positive test results to the infant's parents, guardian, or person in loco parentis.
(4) Subject to the annual adjustment required under this subsection and subject to subsection (6), if the department performs 1 or more of the tests required under subsection (1), the department may charge a fee for the tests of not more than $53.71. The department shall adjust the amount prescribed by this subsection annually by an amount determined by the state treasurer to reflect the cumulative annual percentage change in the Detroit consumer price index. As used in this subsection, "Detroit consumer price index" means the most comprehensive index of consumer prices available for the Detroit area from the bureau of labor statistics of the United States department of labor.
(5) A person who violates this section or a rule promulgated under this part is guilty of a misdemeanor.
(6) The department shall provide for a hardship waiver of the fee authorized under subsection (4) under circumstances found appropriate by the department.
(7) The department shall do all of the following in regard to the blood specimens taken for purposes of conducting the tests required under subsection (1):
(a) By April 1, 2000, develop a schedule for the retention and disposal of the blood specimens used for the tests after the tests are completed. The schedule shall meet at least all of the following requirements:

(i) Be consistent with nationally recognized standards for laboratory accreditation and federal law.
(ii) Require that the disposal be conducted in compliance with section 13811.
(iii) Require that the disposal be conducted in the presence of a witness. For purposes of this subparagraph, the witness may be an individual involved in the disposal or any other individual.
(iv) Require that a written record of the disposal be made and kept, and that the witness required under subparagraph (iii) signs the record.
(b) Allow the blood specimens to be used for medical research during the retention period established under subdivision (a), as long as the medical research is conducted in a manner that preserves the confidentiality of the test subjects and is consistent to protect human subjects from research risks under subpart A of part 46 of subchapter A of title 45 of the code of federal regulations.
(8) The department shall rewrite its pamphlet explaining the requirements of this section when the supply of pamphlets in existence on March 15, 2000 is exhausted. When the department rewrites the explanatory pamphlet, it shall include at least all of the following information in the pamphlet:
(a) The nature and purpose of the testing program required under this section, including, but not limited to, a brief description of each condition or disorder listed in subsection (1).
(b) The purpose and value of the infant's parent, guardian, or person in loco parentis retaining a blood specimen obtained under subsection (9) in a safe place.
(c) The department's schedule for retaining and disposing of blood specimens developed under subsection (7) (a).
(d) That the blood specimens taken for purposes of conducting the tests required under subsection (1) may be used for medical research pursuant to subsection (7) (b).
(9) In addition to the requirements of subsection (1), the health professional described in subsection (1) or the hospital or other facility in which the birth of an infant takes place, or both, may offer to draw an additional blood specimen from the infant. If such an offer is made, it shall be made to the infant's parent, guardian, or person in loco parentis at the time the blood specimens are drawn for purposes of subsection (1). If the infant's parent, guardian, or person in loco parentis accepts the offer of an additional blood specimen, the blood specimen shall be preserved in a manner that does not require special storage conditions or techniques, including, but not limited to, lamination. The health professional or hospital or other facility employee making the offer shall explain to the parent, guardian, or person in loco parentis at the time the offer is made that the additional blood specimen can be used for future identification purposes and should be kept in a safe place. The health professional or hospital or other facility making the offer may charge a fee that is not more than the actual cost of obtaining and preserving the additional blood specimen.

History: 1978, Act 368, Eff. Sept. 30, 1978 ;-- Am. 1986, Act 300, Eff. Mar. 31, 1987 ;-- Am. 1987, Act 14, Imd. Eff. Apr. 14, 1987 ;-- Am. 1988, Act 264, Imd. Eff. July 15, 1988 ;-- Am. 1992, Act 81, Imd. Eff. June 2, 1992 ;-- Am. 1998, Act 88, Imd. Eff. May 13, 1998 ;-- Am. 1999, Act 138, Imd. Eff. Oct. 5, 1999 ;-- Am. 2000, Act 33, Imd. Eff. Mar. 15, 2000 ;-- Am. 2002, Act 691, Eff. Apr. 1, 2003
Popular Name: Act 368
Admin Rule: R 325.1471 et seq. of the Michigan Administrative Code.

Appendix 1 Continued

You can find the law in its entirety at the following link:
http://www.legislature.mi.gov/(li5g3jafe4e5ad55oo2kgxym)/documents/2005-2006/publicact/pdf/2006-PA-0031.pdf

PA 31 2006

STATE OF MICHIGAN
93RD LEGISLATURE
REGULAR SESSION OF 2006

Introduced by Senators George, Hardiman, Allen, Birkholz, Kuipers, Goschka, McManus, Jacobs and Bernero

ENROLLED SENATE BILL No. 794

AN ACT to amend 1978 PA 368, entitled "An act to protect and promote the public health; to codify, revise, consolidate, classify, and add to the laws relating to public health; to provide for the prevention and control of diseases and disabilities; to provide for the classification, administration, regulation, financing, and maintenance of personal, environmental, and other health services and activities; to create or continue, and prescribe the powers and duties of, departments, boards, commissions, councils, committees, task forces, and other agencies; to prescribe the powers and duties of governmental entities and officials; to regulate occupations, facilities, and agencies affecting the public health; to regulate health maintenance organizations and certain third party administrators and insurers; to provide for the imposition of a regulatory fee; to provide for the levy of taxes against certain health facilities or agencies; to promote the efficient and economical delivery of health care services, to provide for the appropriate utilization of health care facilities and services, and to provide for the closure of hospitals or consolidation of hospitals or services; to provide for the collection and use of data and information; to provide for the transfer of property; to provide certain immunity from liability; to regulate and prohibit the sale and offering for sale of drug paraphernalia under certain circumstances; to provide for the implementation of federal law; to provide for penalties and remedies; to provide for sanctions for violations of this act and local ordinances; to provide for an appropriation and supplements; to repeal certain acts and parts of acts; to repeal certain parts of this act; and to repeal certain parts of this act on specific dates," (MCL 333.1101 to 333.25211) by adding sections 5430 and 5432.

The People of the State of Michigan enact:

Sec. 5430. (1) The newborn screening quality assurance advisory committee is created in the department. The newborn screening quality assurance advisory committee shall consist of 10 members and be appointed by the department as follows:

(a) One individual representing a Michigan nonprofit health care corporation.
(b) One individual representing the Michigan health and hospital association.
(c) One individual representing the Michigan state medical society.
(d) One individual representing the Michigan osteopathic association.
(e) One individual representing the department's medical services administration.
(f) One individual representing the department's public health administration.
(g) One individual who is a neonatologist with experience and background in newborn screening.
(h) One individual representing health maintenance organizations.
(i) Two individuals representing the general public.

(10)

Act No. 31
Public Acts of 2006
Approved by the Governor
February 22, 2006

Appendix 1 Continued

Filed with the Secretary of State
February 23, 2006
EFFECTIVE DATE: February 23, 2006
2

(2) The newborn screening quality assurance advisory committee shall meet annually to review the list of newborn screening tests required under section 5431 and under department rules, regulations, and guidelines. The newborn screening quality assurance advisory committee shall, on an annual basis, submit a written report to the department regarding the appropriateness of the existing list of required newborn screening tests. The newborn screening quality assurance advisory committee shall also include in the report recommendations to revise the list to include additional newborn screening tests that are nationally recognized in the scientific literature or national standards for conditions that can be ameliorated or treated if identified by a newborn screening test and to remove certain tests that are no longer supported in the scientific literature or national standard as being effective for ameliorating or treating conditions that can be identified by newborn screening.

(3) The newborn screening quality assurance advisory committee shall conduct a financial review of any recommended changes to the list of newborn screening tests and shall include in the written report required under subsection (2) a recommendation for the increase or decrease in the amount charged pursuant to section 5431 for newborn screening tests. The recommended change shall not exceed any net change in the amount of the actual cost of any proposed additional tests and follow-up minus savings from any proposed deleted tests and follow-up.

(4) Within 30 days after the department has received the report required under subsection (2), the department may approve or reject the recommendations of the newborn screening quality assurance advisory committee. If the department does not reject the recommendations or fails to act within the 30 days, then the recommendations shall be forwarded to the standing committees in the senate and house of representatives that consider issues pertaining to public health for approval.

(5) Within 45 days after the recommendations are forwarded and received, the legislature shall approve or reject those recommendations without amendment by concurrent resolution adopted by both standing committees of the senate and house of representatives that consider issues pertaining to public health and both houses of the legislature by recorded vote. If the proposed recommendations are not submitted on a legislative session day, the 45 days commence on the first legislative session day after the recommendations are submitted. The 45 days shall include not less than 9 legislative session days. If the recommendations are not rejected within the 45-day period, the recommendations shall be considered approved, shall be adopted by the department, and shall take effect 6 months after the recommendations are adopted by both houses of the legislature or considered approved as provided under this subsection.

Sec. 5432. If a health professional in charge of the care of a newborn infant or, if none, the health professional in charge at the birth of an infant, the hospital, the health department, or other facility administers or causes to be administered to the infant a hearing test and screening, then that person or facility shall report to the department, on a form as prescribed by the department, the results of all hearing tests and screens conducted on infants who are less than 12 months of age and on children who have been diagnosed with hearing loss and are less than 3 years of age. The report shall include the type, degree, and symmetry of the diagnosis, along with where and when the diagnosis was made.

This act is ordered to take immediate effect.

Secretary of the Senate
Clerk of the House of Representatives
Approved
Governor

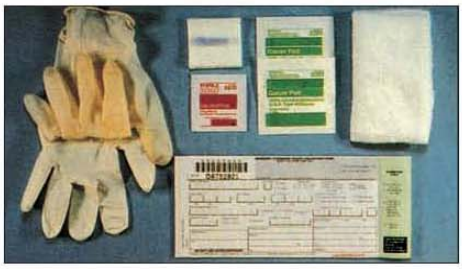
Appendix 2

Blood Specimen Collection and Handling Procedure

Whatman®

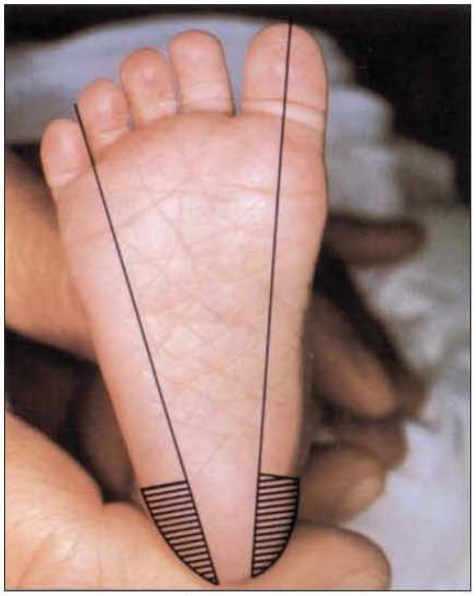
Neonatal Screening

Blood Specimen Collection and Handling Procedure

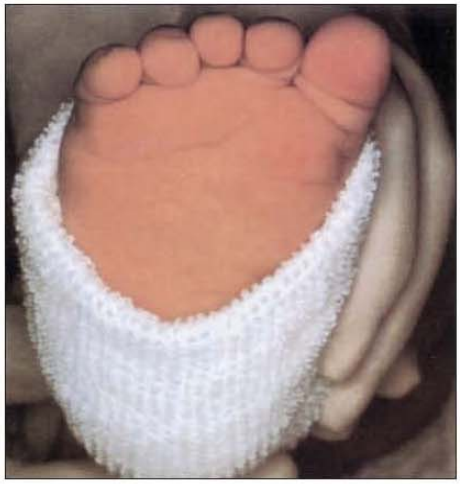


1 Equipment: sterile lancet with tip approximately 2.0 mm – sterile alcohol prep, sterile gauze pads, soft cloth, blood collection form, gloves.

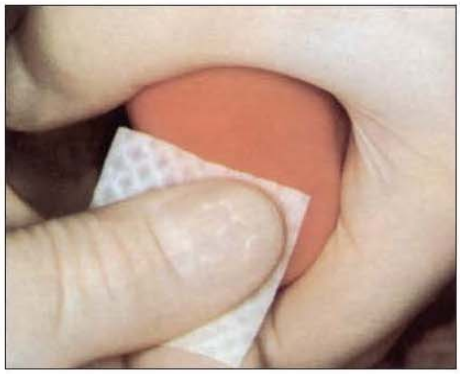
2 Complete ALL information. Do not contaminate filter paper circles by allowing the circles to come into contact with spillage or by touching before or after blood collection. Keep "SUBMITTER COPY" if applicable.



3 Hatched area (▨) indicates safe areas for puncture site.



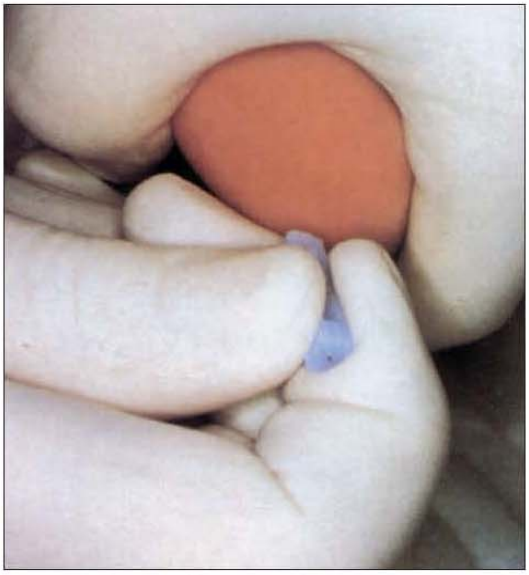
4 Warm site with soft cloth, moistened with warmwater up to 41°C, for three to five minutes.



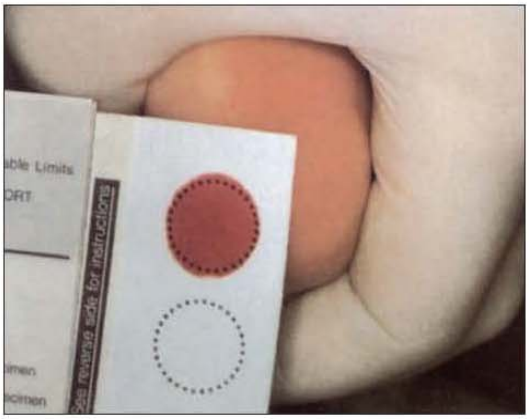
5 Cleanse site with alcohol prep. Wipe DRY with sterile gauze pad.

Over

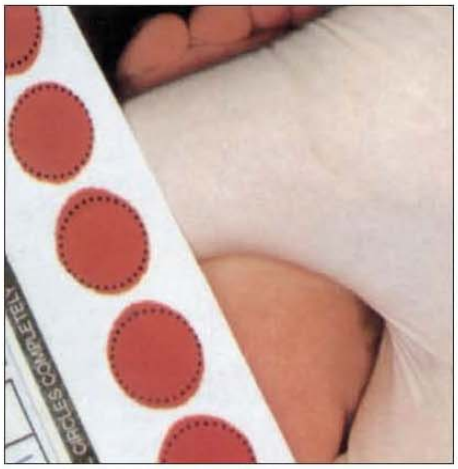
Appendix 2 Continued



6 Puncture heel. Wipe away first blood drop with sterile gauze pad. Allow another LARGE blood drop to form.



7 Lightly touch filter paper to LARGE blood drop. Allow blood to soak through and completely fill circle with SINGLE application of LARGE blood drop. (To enhance blood flow, VERY GENTLE intermittent pressure may be applied to the area surrounding the puncture site). Apply blood to one side of filter paper only.



8 Fill remaining circles in the same manner as step 7, with successive blood drops. If blood flow is diminished, repeat steps 5 through 7. Care of skin puncture site should be consistent with your institution's procedures.

9 Dry blood spots on a dry, clean, flat, non-absorbent surface for a minimum of four hours.

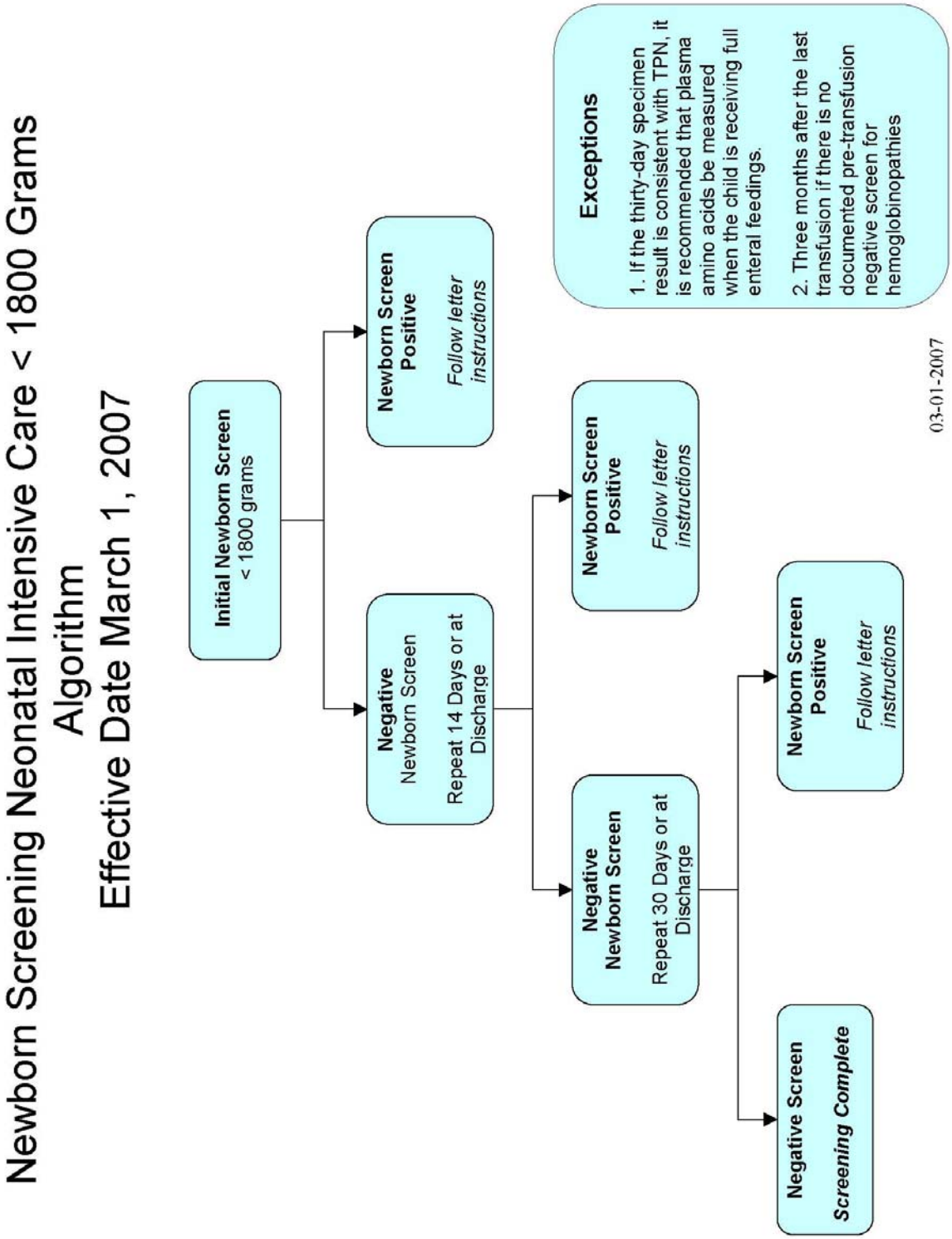
10 Mail completed form to testing laboratory within 24 hours of collection.

North America – Whatman Inc. • Tel: 1-800-WHATMAN • Tel: 1-973-245-8300 • Fax: 1-973-245-8329 • E-mail: info@whatman.com
Europe – Whatman International Ltd • Tel: +44 (0) 1622 676670 • Fax: +44 (0) 1622 677011 • E-mail: information@whatman.com
Whatman GmbH • Tel: +49 (0) 5561 791 0 • Fax: +49 (0) 5561 791 536 • E-mail: information@whatman.com
Japan – Whatman Japan KK • Tel: +81 (0) 3 5215 1242 • Fax: +81 (0) 3 5215 1246 • E-mail: japaninfo@whatman.com
Asia Pacific – Whatman Asia Pacific Pte Ltd • Tel: +65 6534 0138 • Fax: +65 6534 2166 • E-mail: wap@whatman.com

51684(US) S9036-812(EU) 09/05

Appendix 3

Newborn Screening NICU Algorithm



Appendix 4

Disorders List

The Newborn Screening Laboratory screens all Michigan infants for over forty disorders.

Amino Acid Disorders:
Phenylketonuria (PKU)
Benign hyperphenylalaninemia (H-PHE)
Biopterin cofactor biosysnthesis (BIOPT (BS))
Defects of biopterin cofactor regeneration (BIOPT(Reg))
Maple syrup disease (MSUD)
Homocystinuria
Hypermethioninemia (HCY/MET)
Citrullinemia (CIT)
Citrullinemia Type II (CIT II)
Argininosuccinic acidemia (ASA)
Tyrosinemia Type I (TYR I)
Argininemia (ARG)

Fatty Acid Oxidation Disorders:
Carnitine:acylcarnitine translocase deficiency (CACT)
Carnitine palmitoyltransferase II defeficiency (CPT II)
Carnitine uptake defect (CUD)
Carnitine palmitoyltransferase I def. (liver) (CPT 1A)
Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
Glutaric academia type II (GA II)
Med.-chain acyl-CoA dehydrogenase deficiency (MCAD)
Long-chain L-3-OH acyl-CoA de hydrogenase deficiency (LCHAD)
Trifunctional protein def.(LCHAD/TFP)
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
Med.-chain ketoacyl-CoA thiolase deficiency (MCKAT)
Med./short-chain L-3-OH acyl-CoA dehydrogenase deficiency (M/SCHAD)
Dienoyl-CoA reductase deficiency (DE RED)

Organic Acid Disorders:
Isovaleric academia (IVA)
2-Methyl butyryrl-CoA dehydrogenase deficiency (2MBG)
3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
3-OH 3-CH3 glutaric aciduria (HMG)
3-Methylglutaconic aciduria (3MGA)
Beta-ketothiolase deficiency (BKT)
Glutaric acidemia type I (GA I)
Propionic academia (PA)
Methylmalonic acidemia (mutase deficiency) (MUT)
Methylmalonic acidemia (Cbl A,B)
Methylmalonic acidemia (Cbl C,D)
Multiple carboxylase deficiency (MCD)
2-Methyl 3 hydroxy butyric aciduria (2M3HBA)
Malonic acidemia (MAL)
Isobutyryl-CoA dehydrogenase deficiency (IBG)

Endocrine Disorders:
Congenital Adrenal Hyperplasia (CAH)
Congenital Hypothyroidism (CH)

Enzyme Disorders:
Galactosemia (GALT)
Biotinidase Deficiency (BIOT)

Hemoglobinopathies:
Sickle cell anemia (Hb SS)
Hb S/C Disease (Hb S/C)
Hb S/Beta-thalassemia (Hb S/Beta-Th)
Variant Hb-pathies (Var Hb)

Appendix 5

Hearing Screening Contact Information

Michigan Department of Community Health Hearing Program
Early Hearing Screening Detection and Intervention
Division of Family and Community Health
109 E. Michigan Avenue
P.O. Box 30195
Lansing, MI 48909

Name	Position	Phone	E-mail
Michelle Garcia, Au.D, CCC-A	Follow-up consultant	(517) 335-8878	garciam@michigan.gov
Lorie Lang, MA, CCC-A	Audiology Consultant	(517) 335-9125	langlo@michigan.gov
Kylie Sharp	Parent Consultant	(517) 335-8273	sharpk@michigan.gov
Debby Behringer, RN, MSN	Community Development Consultant	(517) 335-8875	behringerd@michigan.gov
Erin Estrada, BA	Data Analyst	(517) 335-8916	estradae@michigan.gov
Elizabeth Willcutt	General Office Assistance	(517) 335-8955	Willcutte1@michigan.gov

_____Michigan EHDI Web page_____

http://www.michigan.gov/ EHDI

National Newborn Hearing Websites

CDC EHDI
Website: http://www.CDC.GOV/ncbddd/ehdi
E-mail: ehdi@cdc.gov

Marion Downs National Center for Infant Hearing
Website: http://www.colorado.edu/slhs/mdnc
E-mail: mdnc@colorado.edu

National Center for Hearing Assessment and Management
Website: http://www.infanthearing.org
National Institute on Deafness and Other Communication Disorders
http://www.nih.gov/nidcd.recomnd.htm
http://www.nih.gov/nidcd/homepage.htm

www.aap.org
www.asha.org
www.audiology.org
www.handsandvoices.org

Appendix 6

Medical Management Centers

Sickle Cell – *Hemoglobinopathies*

SICKLE CELL ASSOC. OF AMERICA, MICHIGAN CHAPTER, INC.
18516 James Couzens
Detroit, MI 48235
(P) 313-864-4406 (F) 313-864-9980

Social work and Counseling sites (ONLY) CALL Sickle Cell Assoc. for referral

Urban League of Grand Rapids
745 Eastern Ave., S.E.
Grand Rapids, MI 48503

United Way of Oakland County
50 Wayne St.
Pontiac, MI 48342

Northside Opportunity For Ownership
612 N. Park St.
Kalamazoo, MI 49441

Lansing Mt. Zion Missionary
1314 Ballard Street
Lansing, MI 48906

Berrien County Health Department
769 Pipestone St.
Benton Harbor, MI 49023

Urban League of Flint
5005 Cloverlawn
Flint, MI 48505

Family Service Workforce Development Center
1516 Peck St.
Muskegon, MI 49441

Saginaw Centre Development
310 Johnston St. Rm 299
Saginaw, MI 48607

CHILDREN'S HOSPITAL OF MICHIGAN
3901 Beaubien Blvd.
Detroit, MI 48201-2192
(P) 313-745-5613 (F) 313-745-5237

Metabolic – *Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, Other Disorders - Galactosemia and Biotinidase Deficiency*

CHILDREN'S HOSPITAL OF MICHIGAN
3901 Beaubien Blvd.
Detroit, MI 48201-2192
(P) 313-745-4513 (F) 313-745-4827

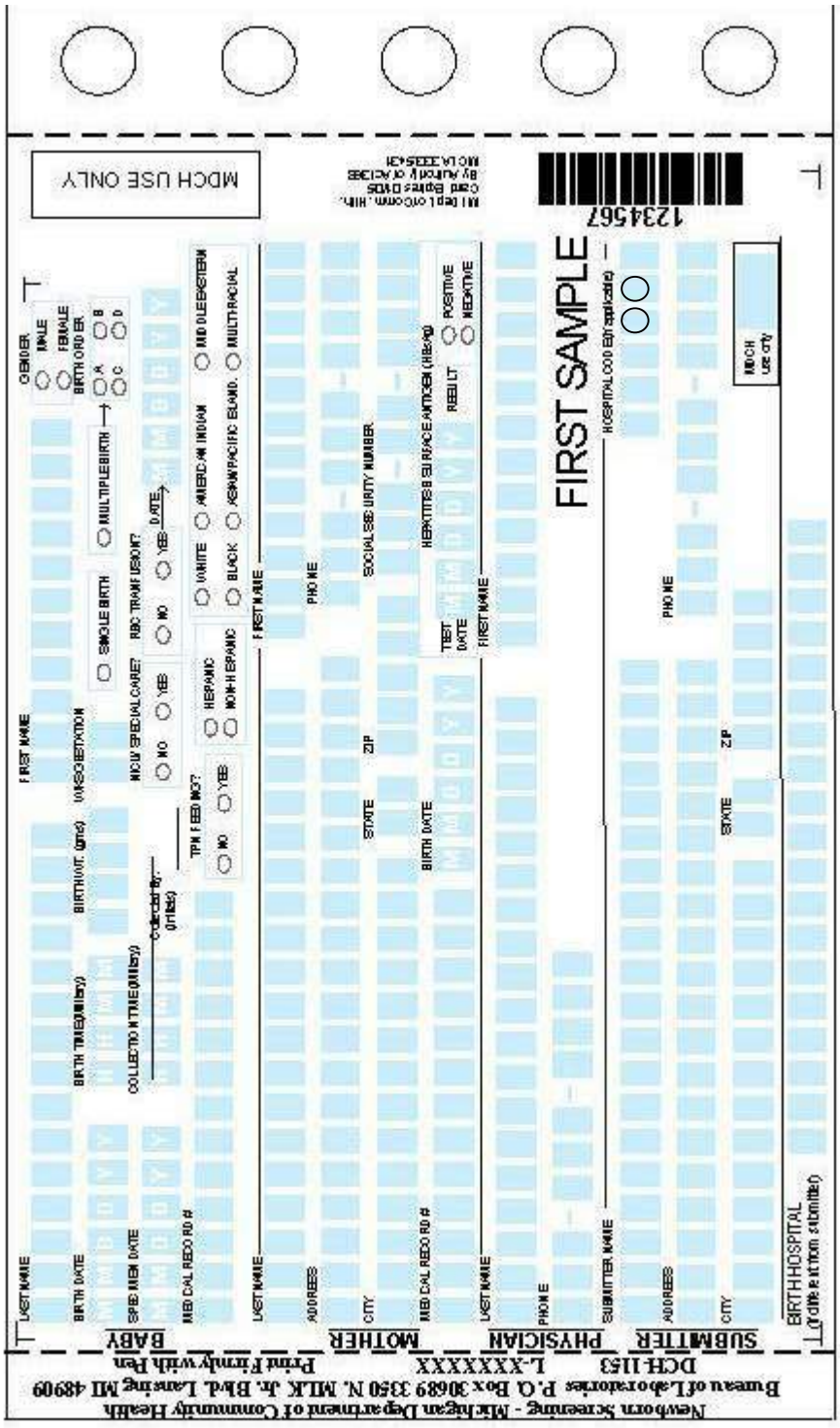
Endocrine – *Congenital Hypothyroidism (CH), Congenital Adrenal Hyperplasia (CAH)*

PEDIATRIC ENDOCRINE FOLLOW-UP CLINIC
University of Michigan Health System
Department of Pediatrics
1500 E. Medical Ctr. Dr.
D1225 MPB, Box 0718
Ann Arbor, MI 48109-0718
(P) 734-647-8938

CAH - CENTER OF EXCELLENCE
University of Michigan Health System
Department of Pediatrics
1500 E. Medical Ctr. Dr.
D1225 MPB, Box 0718
Ann Arbor, MI 48109-0718
(P) 734-647-8938

Appendix 7

Specimen Card



Appendix 8

HIPAA Privacy Rule

The HIPAA Privacy Rule recognizes the need for public health programs to access protected health information (PHI) to conduct public health activities to prevent or control disease, injury or disability. The Privacy Rule* expressly permits release of PHI relating to newborn screening, without individual authorization, from a covered entity to state public health departments or agencies contacted, by public health departments, to provide newborn screening follow-up.

* http://www.cdc.gov/mmwr/preview/mmwrhtml/m2e411a1.htm

Appendix 9

Parental Refusal for Newborn Screening

We (I)_____, the parent(s) or guardian(s) of

_____Birth date_____
object to the requirement that our (my) child be tested to determine the presence of a variety of disorders , including, Phenylketonuria (PKU), Congenital Hypothyroidism, Galactosemia, Maple Syrup Urine Disease (MSUD), Biotinidase Deficiency (BD), Sickle Cell Anemia and other Hemoglobinopathies, Congenital Adrenal Hyperplasia (CAH), Medium Chain Acyl Co-A Dehydrogenase Deficiency (MCAD), Homocystinuria, Citrullinemia, and Argininosuccinic Aciduria (ASA).

We (I) have been fully informed and fully understand the possible devastating consequences to our (my) child's health resulting from undetected and untreated disorders, including, Phenylketonuria (PKU), Congenital Hypothyroidism, Galactosemia, Maple Syrup Urine Disease (MSUD), Biotinidase Deficiency (BD), Sickle Cell Anemia and other Hemoglobinopathies, Congenital Adrenal Hyperplasia (CAH), Medium Chain Acyl Co-A Dehydrogenase Deficiency (MCAD), Homocystinuria, Citrullinemia, and Argininosuccinic Aciduria (ASA) including severe mental and/or physical impairment or death. We (I) have had the opportunity to review A First Step to Your Baby's Health.

Nevertheless, we (I) choose not to have _____ tested for a variety of disorders, including, Phenylketonuria (PKU), Congenital Hypothyroidism, Galactosemia, Maple Syrup Urine Disease (MSUD), Biotinidase Deficiency (BD), Sickle Cell Anemia and other Hemoglobinopathies, Congenital Adrenal Hyperplasia (CAH), Medium Chain Acyl Co-A Dehydrogenase Deficiency (MCAD), Homocystinuria, Citrullinemia, and Argininosuccinic Aciduria (ASA).

Therefore, we (I) release the Michigan Department of Community Health, the hospital of birth and the person responsible for collection of the specimen of responsibility for screening our (my) child for the above-mentioned diseases. Furthermore, we (I) release and hold the Michigan Department of Community Health, the hospital of birth and the person responsible for collection of the specimen harmless for any injury, illness, and/or sequelae which may result to our (my) child as the result of our (my) refusal to consent to the above tests.

_____ _____
Signature of parents(s) or guardian(s) date

_____ _____
Signature of parents(s) or guardian(s) date

_____ _____
Signature of witness date

_____ _____
Signature of witness date

cc: Michigan Department of Community Health
Newborn Screening Program
201 Townsend Street
P. O. Box 30195
Lansing, MI 48909

Appendix 10

Newborn Screening NICU Fact Sheet

Michigan Department of Community Health

Newborn Screening for NICU Infants < 1800 Grams Provider Fact Sheet

Newborn screening and premature infants
Newborn Screening is an important part of infant health maintenance. However, like so many other programs designed primarily for the healthy term baby, newborn screening of the premature, low birth weight, and ill infants is not a simple or straightforward process. The neonates' immaturity and the necessary therapeutic interventions combine to interfere with both the collection of samples and the interpretation of newborn screening results.

Why should premature infants be screened differently?
Premature infants should be screened differently to minimize both the false positive and false negative results in these small babies. Collecting three specimens from each infant, and viewing the results together, will give a clearer picture of the neonate's risk for the disorders included in Michigan's screening panel.

How should the specimens be collected?
Specimens should be collected on the blue screening cards at 24-36 hours after birth, unless the infant receives blood. In this case, obtain the specimen prior to blood administration including ECHMO (Extracorporeal Membrane Oxygenation). Repeat specimens are obtained on pink cards at 14 and 30 days of age or upon discharge if discharge is prior to 14 or 30 days of age. Ordering all three screens upon the infant's admission to the NICU will be most efficient. If the baby goes home after the 2nd specimen, then that is the last specimen.

Why obtain specimen before transfusion?
If the infant requires transfusion before 24 hours of age, collect the initial specimen pre-transfusion and the next specimen at 14 and 30 days of age or upon discharge. A pre-transfusion specimen is essential for detection of galactosemia, sickle cell disease, and biotinidase deficiency. If the infant receives a blood transfusion before the screen is collected, the newborn screen must be repeated 90 days post-transfusion. Results from a post transfused specimen are not valid and may represent a false negative.

Are these screens done differently than regular newborn screens?
No. The laboratory testing is the same. Clinicians will still be notified of all abnormal results.

Are the reports different?
The report format is the same for all newborns except as noted below. Please follow instructions on the reports in obtaining repeats when requested.

The following situations are reported differently for infants in the NICU:

- If the initial screen for congenital adrenal hyperplasia (CAH) is positive, the report will suggest clinical evaluation of the infant and a repeat screen at 14 days of age. Positive results on repeat screens will be treated the same way as positive results in other babies.
- If the amino acid pattern is consistent with total parenteral nutrition (TPN) on the initial or 14 day sample, no special action will be recommended; the next screening sample will simply be requested. Only if the result is consistent with TPN on the 30 day specimen is the request made to measure plasma amino acids when the child is receiving full enteral feedings.

Any questions about requests for repeats or infant status in relationship to testing can be answered by medical management centers.

Where can I get additional information?

- Newborn Screening NICU Provider Manual for Michigan is available on-line at: http://www.michigan.gov/newbornscreening Hard copy versions of the manual are provided to Michigan's NICU coordinators.
- The staff of the Newborn Screening Program at the Michigan Department of Community Health is available to answer your questions at 1-866-673-9939.

Michigan would like to acknowledge and thank Minnesota Department of Health staff Abbie Abboud and Beth-Ann Bloom, for their creation of this document and willingness to share it for the benefit of Michigan's children.

TJA 1/26/2007